

FIG. 1 RECEIVED

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1 TTGGTAGAACGGAAACGGCGGCCTTCGGCCGGCTCCGGCGCTCCTGGTC
61 TCGGCGGGCCTCCCCGCCCTTCGTCTCGTCCTCTCCCCCTGCCAGCCC
121 CTCCGGCCGCGCCAACCCGCGCCTCCCCGCTCGGCGCCGTGCGTCCCCGCC
181 GCGTCTCCTGGCGGCCGGCTCCGGCTGTCCCCGCCGGCGTGCAGCCGGTATG
SCA2-A
241 GGCCCCCTCACCATGTCGCTGAAGCCCCAGCAGCAGCAGCAGCAGCAGCAACAGCAGC
SCA2-B
301 AGCAGCAACAGCAGCAGCAGCAGCAGCAGCAGCAGGCCGCCGCCGGCTGCCAATGTCCGCA
361 AGCCCCGGCGGCAGCGGCCTTCTAGCGTCGCCGCCGCCGCCCTCGCCGTCTCGTCCT
421 CGGTCTCCTCGTCCTCGGCCACGGCTCCCTCCTCGGTGGTCGGCGACCTCCGGCGCG
481 GGAGGGCCCGGCCTGGGCAG GTGGGTGTGGCACCCCC

FIG. 2



SAK2041
SAK2047
SAK2054
SAK2065
SAK2066
SAK4781
SAK2963
SAK2966
SAK2937
FSIV-8A
FSIV-9
FSIV-2A
FSII-8
FSII-24
FSIII-27

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 A C G T

52 →

36 →

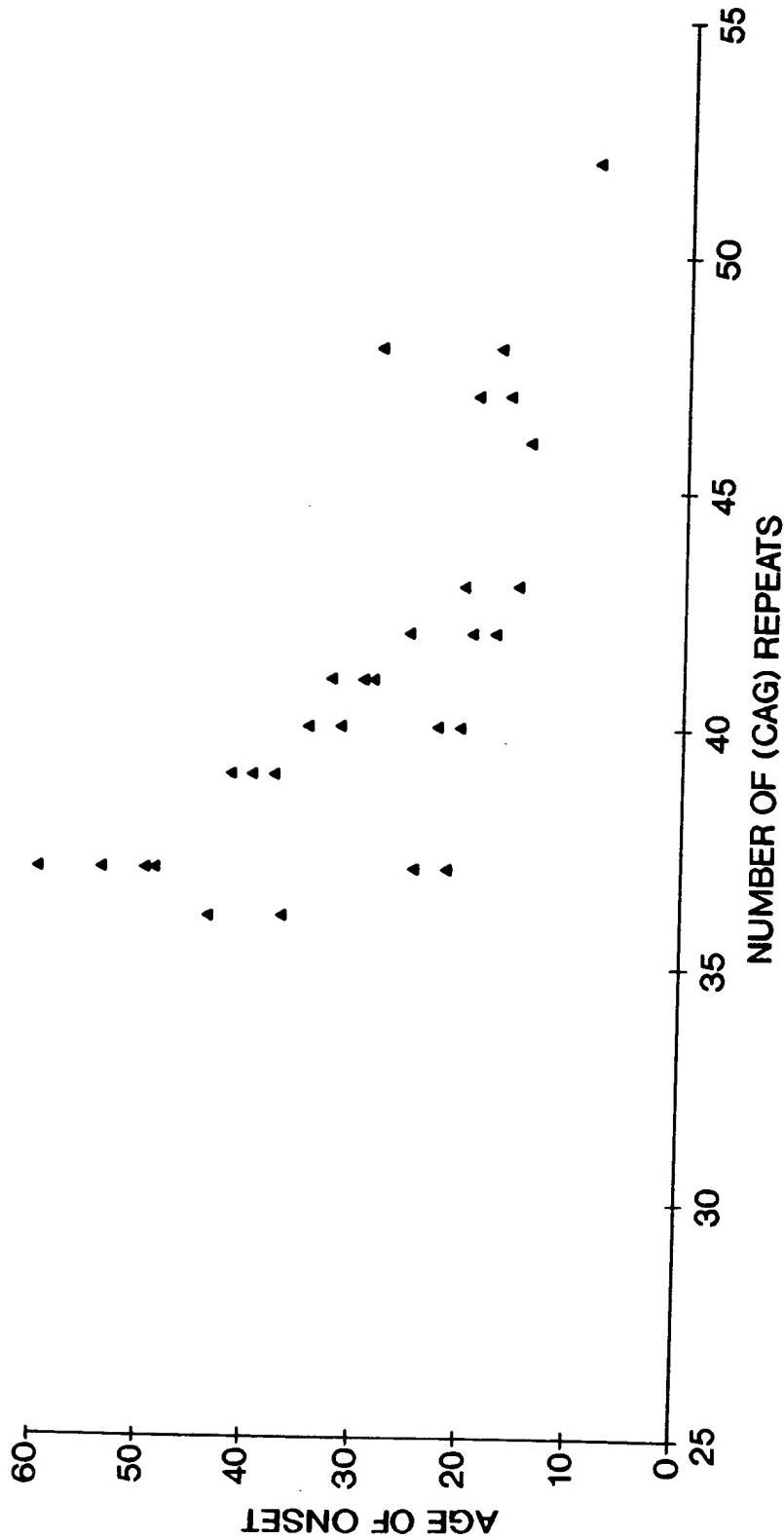
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FIG. 3



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FIG. 4



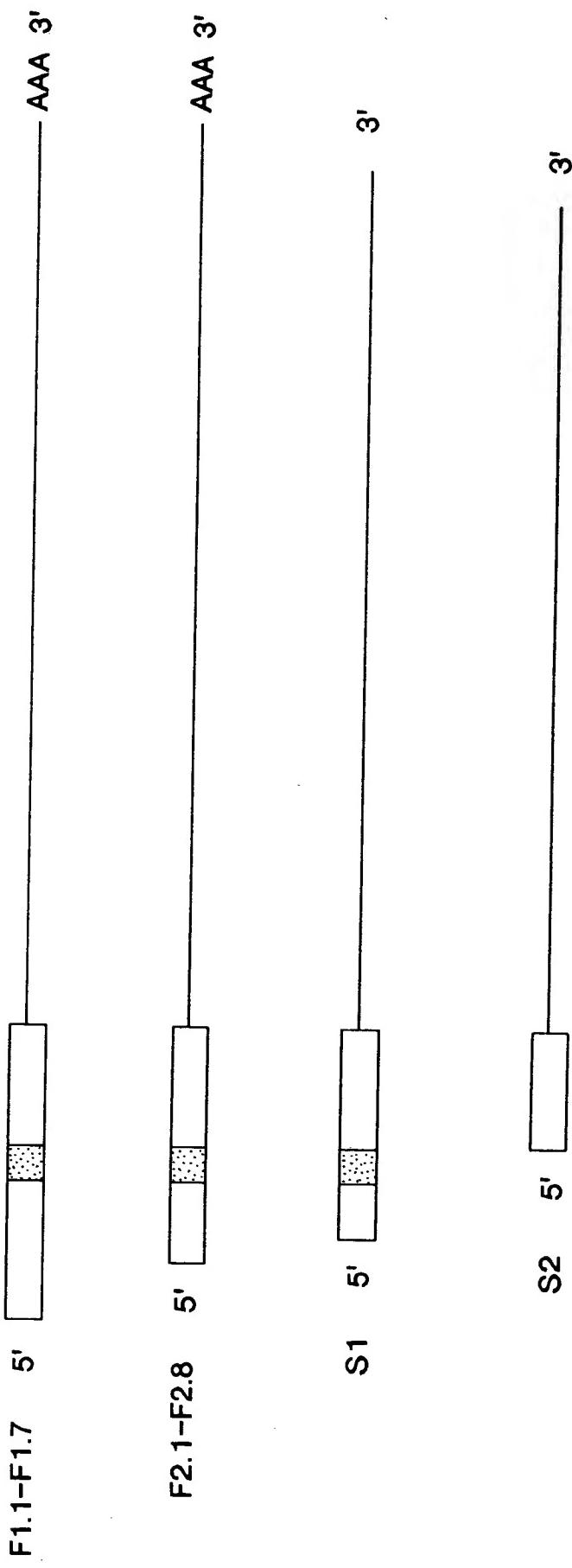


FIG. 5

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FIG. 6A

721 CAGCCGGCGGGCTGCCAATGTCGGCAAGCCCCGGGGCAGGGCTTCTAGGGTCG 780
187 Q P P A A N V R K P G G S G L L A S 206
781 CCCGGCGGGCTTCGGGTCCCTCGTCCCTCGTCCTCGGCCACGGCTCCC 840
207 P A A P S P S S V S S S A T A P 226
841 TCCTCGGTGGTGGCACCTCGGGGAGGGGGGGGGAGGGCTGGCAGAGGTGAAAC 900
227 S S V V A A T S G G G R P G L G R N 246
901 AGTAACAAAGGACTGGCTCAGTCTACGGATTCTTTGATGGAATCTATGCCAATATGAGG 960
247 S N K G L P Q S T I S F D G I Y A N M R 266
961 ATGGTTCATATACTTACATCAGTTGGCTCCAAATGTCAAGTACAAGTGAATAATGGA 1020
267 M V H I L T S V V G S K C E V Q V K N G 286
SCA2-14B
1080
287 G I Y E G V F K T Y S P K C D L V L D A 306
1081 GCACATGAGAAAGTACAGAATCCAGTTCGGGGGAAACGTTGAAAGAAATAATGGAGGT 1140
307 A H E K S T E S S G P K R E E I M E S 326
1141 ATTGTTGTTCAATGTTCAAGACTTGTGTGTTACAGTTAAAGATATGGACTCCAGTTAT 1200
327 I L F K C S D F V V Q F K D M D S S Y 346
1201 GCAAAAGAGATGGCTTTACTGACTCTGTATCAGTGCTAAAGTGAATGGCAAACACAAA 1260
347 A K R D A F T D S A I S A K V N G E H K 366
1261 GAGAAGGACCTGGAGCCCTGGGATGCAGGTAACTCACAGCCAATGGAAACTTGAGGCT 1320
367 E K D L E P W D A G E L T A N E E L E A 386
1321 TTGGAAAAATGACCTATCTATGGATGGATCCAAATGATATGTTCGATAATAATGAAGAA 1380
387 L E N D V S N G W D P N D M F R Y N E E 406
1381 AATTATGGTGTAGTGTCTACGTATGAGCAAGTTATCTTCGTATAACAGTGCCTTAGAA 1440
407 N Y G V V S T Y D S S L S S Y T V P L E 426
1441 AGAGATAACTCAGAAGAAATTTTAACCGGGAAAGCAAGGGCAAACCAGTTAGCAGAAGAA 1500
427 R D N S E E F L K R E A R A N Q L A E E 446

FIG. 6B

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1501 ATTGAGTCAAGTCCCCAGTACAAGCTCGAGTGGCCCTGGAAAATGATGATAGGAGTGAG 1560
447 I E S S A Q Y K A R V A L E N D D R S E 466
1561 GAAGAAAAATAACACAGCAGTTCAAGAGAAATTCCAGTGAACGTTGAGGGCACAGGCATAAAC 1620
467 E E K Y T A V Q R N S S E R E G H S I N 486
1621 ACTAGGGAAAATAAAATAATATTCTCTGGACAAAGAAATAGAGAAGTCATATCCTGGGGA 1680
487 T R E N K Y I P P G Q R N R E V I S W G 506
1681 AGTGGAGACAGAAATTCAACCCGGTATGGGCCAGGCCTGGATGGGCTCCATGCCATCAAAGA 1740
507 S G R Q N S P R M G Q P G S M P S R 526
1741 TCCACTTCTCACACTTCAGATTCAACCCGAATTCTGGTTCAAGACCAAAGAGTAGTTAAT 1800
527 S T S H T S D F N P N S G S D Q R V V N 546
1801 GGAGGGTGTCCCTGGCCATGGCCCATGGCCATTCTCCTCTCGCCCACCTTCTCGCTAC 1860
547 G G V P W P S P C P S P S R P P S R Y 566
1861 CAGTCAGGTCCCACACTCTCTCCACCTCGGGCAGCCACCCCTACACGGGCCCTCCAGG 1920
567 Q S G P N S L P P R A A T P T R P P S R 586
1921 CCCCTCTGGGCCATCCAGAGCCCCATGGCTCACCCCTCTGCTCATGGTTCTCCAGCTCCT 1980
587 P P S R P S R P P S H P S A H G S P A P 606
1981 GTCTCTACTATGCCCTAAACCCATGTTCAAGAAGGGCTCCAAGGGATGTCCTCCAAAGGCC 2040
607 V S T M P K R M S S E G P P R M S P K A 626
2041 CAGCGACATCCTCGAAATCACAGAGTTCTGGCTGGGGGGTTCCATATCCAGTGGCCTA 2100
627 Q R H P R N H R V S A G R G S I S S G L 646
2101 GAATTGTATCCACACCCACCCAGTGAAGGCAGCTACTCCTCCAGTAGCCAAGGACCAGT 2160
647 E F V S H N P P S E A A T P P V A R T S 666
2161 CCCTGGGGAACGGTGGTCACTCAGTGGTCAAGTGGGGTTCCAAGATTATCCCTAAACT 2220
667 P S G G T W S S V V S G V P R L S P K T 686
2221 CATAGACCCAGGTCTCCACAGAACAGTATTGGAAATAACCCCCAGTGGGCCAGTTCTT 2280
687 H R P R S P R Q N S I G N T P S G P V L 706

FIG. 6C

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2281	GCTTCTCCCCAAGCTGTTATTCCAACCTGAAGCTGGTGTGCCATGCCATTCCAGCTGCA	2340
707	A S P Q A G I I P T E A V A M P I P A A	726
2341	TCTCCTACGGCCTGCTAGTCCTGCATCGAACAGAGCTGGTACCCCTTCTAGTGAGGCTAAA	2400
727	S P T P A S P A S N R A V T P S S E A K	746
2401	GATTCCAGGCTTCAAAGATCACAGGGCAGAACACTCTCCTGCAGGGAAATAAAGAAATAATTAAA	2460
747	D S R L Q D Q R Q N S P A G N K E N I K	766
2461	CCCAATGAAACATCACCTAGCTTCTCAAAAGCTGAAAACAAGGTATATCACCAGCTTGT	2520
767	P N E T S P S F S K A E N K G I S P V V	786
2521	TCTGAAACATAGAAAACAGATTGATGATTAAAGAAATTAAAGAAATGATTAGTTAGGTTACAG	2580
787	S E H R K Q I D D L K K F K N D F R L Q	806
2581	CCAAGTTCTACTTCTGAATCTATGGATCAACTAAACAAACAAATAAGAGAGGGAGAAAAA	2640
807	P S S T S E S M D Q L L N K N R E G E K	826
2641	TCAAGAGGATTGTATCAAAGACACAAATTGAAACCAAGTGTAAAGGATTCTCATTTGAAAT	2700
827	S R D L I K D K I E P S A K D S F I E N	846
2701	AGCAGCAGCAACTGTACCAAGTGGCAGGAGCCAAAGGCCAAATAGCCCCCAGGCATTCCCCCTCA	2760
847	S S N C T S G S S K P N S P S I S P S	866
2761	ATACTTAGTAACACGGACACAAGGGGGACCTGAGGTCACTTCCCAGGGTTCAGACT	2820
867	I L S N T E H K R G P E V T S Q G V T	886
2821	TCCAGGCCAGCATGTAACAAAGAGACGATAAGGAAGAGAAAGAACCCAGCTGAG	2880
887	S S P A C K Q E K D D K E E K K D A A E	906
2881	CAAGTTAGGAAATCAACATTGAATCCCAATGCAAAGGAGTCAACCCACAGTTCCTCTCT	2940
907	Q V R K S T L N P N A K E F N P R S F S	926
2941	CAGCCAAAGCCTCTACTACCTCACTTCCGGCCTCAAGCACAAACCTAGGCCATCT	3000
927	Q P K P S T T P S P R P Q A Q P S P S	946
3001	ATGGTGGGTCAATCAACAGCCAACCTCCAGTTTATACTCAGGCTGTGTTGCAACAAAT	3060
947	M V G H Q Q P T P V Y T Q P V C F A P N	966
3061	ATGATGTTATCCAGTCCCAGTGAGGCCAGGGCGTGCACCTTATAACCCAATACCTATGACG	3120
967	M M Y P V P V S P G V Q P L Y P I P M T	986

FIG. 6D

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3121 CCCATGCCAAGTGAATCAAGCCAAGACATATAAGCAGTACCAAATATGCCAACAGGGG 3180
987 P M P V N Q A K T Y R A V P N M P Q Q R 1006
3181 CAAGACCAGCATCATCAGAGTGCCATGATGCCACCCAGCGTCAGCGGGCCACCGATT 3240
1007 Q D Q H H Q S A M M H P A S A A G P P I 1026
3241 GCAGGCCACCCCCACCCAGCTTAACTCCACGGCAATAATGTTGCCTACAGTCCTCAGCAGTTCCCA 3300
1027 A A T P P A Y S T Q Y V A Y S P Q Q F P 1046
3301 AATCAGCCCCCTTGTTCAGCATGTCACATATCAGTCTCAGCATGTCATGTCATAGT 3360
1047 N Q P L V Q H V P H Y Q S Q H P H V Y S 1066
3361 CCTGTAATAACGGGTAATGCTAGAAATGATGGCACCAACACAGCCCAGGCCAGGTGTTA 3420
1067 P V I Q G N A R W M A P P T H A Q P G L 1086
3421 GTATCTTCTTCAGCAACTCAAGTACGGGCTCATGAGCAGACCCATGGCATGTGCATGT 3480
1087 V S S S A T Q Y G A H E Q T H A M Y A C 1106
3481 CCCAAATTACCATACAAGGAGACAAGCCCTTCTACTTTGCATTTCACGGGC 3540
1107 P K L P Y N K E T S P S F Y F A I S T G 1126
3541 TCCCTTGCTCAGCAAGTATGGCACCCCTAACGGCTACCTGCACCCACATACTCCACCCCT 3600
1127 S L A Q Q Y A H P N A T L H P H T P H P 1146
3601 CAGCCTTCAAGCTACCCCCACTGGACAGCAGCAAAAGCCAACATGGTGGAAAGTCATCCTGCA 3660
1147 Q P S A T P T G Q Q S Q H G G S H P A 1166
3661 CCCAGTCCCTGTTCAAGCACCATTAGGCCACGGCCGGCCAGGCTCTCCATCTGGCCAGTCCA 3720

FIG. 6E

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1167	P	S	P	V	Q	H	H	Q	H	Q	A	A	Q	A	L	H	L	A	S	P
3721	CAGCAGGCACTGCAGCCATTACCCACGGGGCTTGGGCCAACTCCACCCCTCCATGACACCT	3780																		
1187	Q	Q	S	A	I	Y	H	A	G	L	A	P	T	P	P	S	M	T	P	
3781	GCCTCCAACACGGCAGTCGCCAACAGAATAAGTTCCCAGGCCAACAGACTGTCTTTACG	3840																		
1207	A	S	N	T	Q	S	P	Q	N	S	F	P	A	A	Q	Q	T	V	F	T
3841	ATCCATCCTTCTCACGTTCAAGCGGTATAACCAACCCACATGGCCCCACATGGCTTACCT	3900																		
1227	I	H	P	S	H	V	Q	P	A	Y	T	N	P	P	H	M	A	H	V	P
3901	CAGGCTCATGTACAGTCAGGAATGGTTCCCTCTCATCCAAACTGCCATGCCAAATGATG	3960																		
1247	Q	A	H	V	Q	S	G	M	V	P	P	S	H	P	T	A	H	A	P	M
3961	CTAATGACGACACGCCAACCGGGGTCCCCAGGGCCCTGGCTCAAAGTGCACACTACAG	4020																		
1267	L	M	T	Q	P	P	G	G	P	Q	A	A	L	A	Q	S	A	L	Q	
4021	CCCATTCCAGTCTGACAACAGGCATTTCCCTATATGACGGACCCCTTCAGTACAAGCC	4080																		
1287	P	I	P	V	S	T	T	A	H	F	P	Y	M	T	H	P	S	V	Q	A
4081	CACCAACAGCAGCTTAAGGCTGCCCTGGAGGAACCGAAAGGCCAATTCCCTCCCT	4140																		
1307	H	H	Q	Q	Q	L	*													
4141	CCTTCTACTGCTTCTACCAACTGGAAAGCACAGAAAACCTAGAAATTTCATTATTGTTT	4200																		
4201	TAAAATATAATATGTTGATTCTGTAAACATCCAATAGGAATGCTAACAGTTCACTTGCAG	4260																		
4261	TGGAAAGATACTTGGACCGAGTAGAGGCATTAGGAACCTTGGGGCTATTCATAATTCCA	4320																		
4321	TATGCTGTTCAAGACTCCGGTACCCAGCTCTGCTTGGCGAACCTGAAGTTATT	4380																		
4381	ATTTTTTATAACCCTTGAAAGTCATGAACACATCAGCTAGCAAAAGAAGTAACAAAGAGT	4440																		
4441	GATTCCTGCTGCTTAACTGCTAAAAAAAA	4481																		

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FIG. 6F

	Ataxin-2	Ataxin-2	A2RP	Consensus	Ataxin-2	Ataxin-2	A2RP	Consensus
1	VYGPLTMSLK	PQQQQQQQQQ	QQQQQQQQQQ	QQQQPPAAAN	VRKPGGSGLL			
	HEGPLTMSLK	PQPQ	PPAPAT	GRKPGG	GLL	
	...	LA	PQPPPQQHQ	ER
			L-	PQ	---	---	---	---

					100
51	Ataxin-2	ASPAAPSPS	SSSVSSSAT	APSSVVA...	ATSGGGRPGL
	MOUSE Ataxin-2	SSPGAAP.AS	AAVTSAVVP	APAAPVASSS	AAAGGGRPGL
	A2RP	...PGAAAIGS	A...
	Consensus	-P-AA--S	- - - - -	- - - - -	- - - - -

	Ataxin-2	Mouse Ataxin-2	A2RP	Consensus	101	150
PQSTISFDGI	YANVRMVHIL	TSVVGSKCEV	QVKNGGIYEG	VFKTYSPKCD		
PQPTISFDGI	YANVRMVHIL	TSVVGSKCEV	QVKNGGIYEG	VFKTYSPKCD		
PQSPV.FEGV	YNNSRMLHFL	TAVVGSTCDV	KVKNGTTYEG	IFKTLSSKFE		
PO---F-G-	Y-N-RM-H-L	T-VVGS-C-V	-VKNG--YEG	-FKT-S-K--		

Ataxin-2	LVLDAAHEKS	TESSSGPKRE	EIMESILFKC	SDFVVVQFKD	MDSSYYAKRDA
Mouse Ataxin-2	LVLDAAHEKS	TESSSGPKRE	EIMESVLFKC	SDFVVVQFKD	TDSSYYARRDA
A2RP	LAVDAVHRKA	SEPAGGPRRE	DIVDTMVFKP	SDVMLVHFRN	VDFNYATKDK
Consensus	L--DA-H-K-	-E---GP-RE	-I-----FK-	SD---V-F--	-D--YA--D-
					200



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FIG. 7A

201	Ataxin-2	FTDSAIS..A KVNGEHKEKD LEPWDAGELT ANEELEALEN DVSN	GWD PND
	Mouse Ataxin-2	FTDSALS..A KVNGEHKEKD LEPWDAGELT ASEELE.LEN DVSN	GWD PND
	A2RP	FTDSAIAMNS KVNGEHKEKV LQRWEGGD.S NSDDYD.LES DMSN	GWD PNE
	Consensus	FTDSA----- KVNGEHKEK-L--W--G--- -----LE-	D-SNGWDPN-

Ataxin-2	MFRYNEENYG	WVSTYDSSLS	SYTVPLERDN	SEEFLKREAR	ANQLAEEIES
Mouse Ataxin-2	MFRYNEENYG	WVSTYDSSLS	SYTVPLERDN	SEEFLKREAR	ANQLAEEIES
A2RP	MFKFNEENYG	VKTTYDSSLS	SYTVPLEKDN	SEEFRQRELRL	AAQLAREIES
Consensus	MF--NEENYG	V--TYDSSLS	SYTVPLE-DN	SEEF--RE-R	A-QLA-EIES

Ataxin-2	SAQYKARVAL	ENDD . RSEEE	KYTAVQRNSS	EREGHSINTR	ENKYI PPPGQR
Mousse Ataxin-2	SAQYKARVAL	ENDD . RSEEE	KYTAVQRNCS	DREGHGPNTIR	DNKYI PPPGQR
A2RP	SPQYRLRIAM	ENDDGRTEEE	KHSAVQRQGS	GRESPLASR	EGKYI P . . .
Consensus	S - QY - - R - A -	ENDD - R - EEE	K - - AVQR - - S	- RE - - - - R	- - KYIP - - -

351	NR	..
Ataxin-2	NR	--
Mouse Ataxin-2	NR	
A2RP	Consensus	

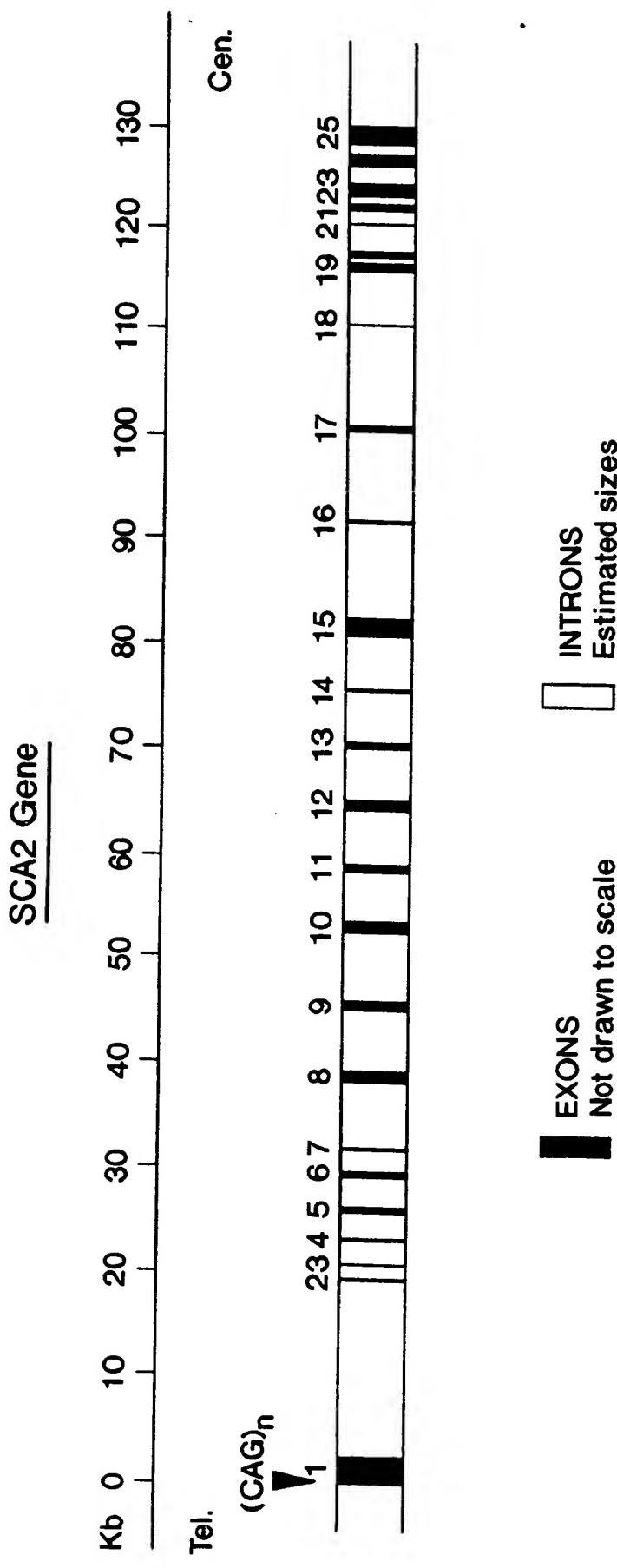


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FIG. 7B



- Largest exon: exon 1, 928 bps; contains CAG repeat
- Largest intron: intron 1 with approximately 15 Kbps
- Smallest exon: exon 2, 37 bps
- Exon sizes:
 - $8 < 100$ bps
 - $100 \text{ bps} < 12 < 200$ bps
 - $200 \text{ bps} < 4 < 400$ bps
 - $400 \text{ bps} < 1$
- Known intron sizes:
 - intron 2 : 1.6 Kb
 - intron 19: 0.3 Kb
 - intron 22: 1.0 Kb
 - intron 24: 1.6 Kb

FIG. 8

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